

PARENT INFORMATION FOR PRESUMPTIVE POSITIVE newborn screening results for CONGENITAL ADRENAL HYPERPLASIA (CAH)



When your baby was born a small sample of blood was taken from your baby's heel to test for certain diseases. These diseases usually can not be found simply by looking at your baby, so that is why the blood test is so important.

The results of your baby's screening test were "presumptive positive" for a disorder called congenital adrenal hyperplasia (a-DREE-nuhl HIGH-per-PLAY-zia). It is important to remember this is only a screening test. A new specimen will be collected from your baby and a new test will be done to help diagnose your baby, or to rule out this disorder.

? What does a "positive" screen mean?

A small percent of all babies screened will be identified as "positive" on screening, but are later found to not have the disorder. This is just a "screening" test and further testing should be done to determine if your child has this disorder, or to rule it out as a "false positive".

☞ What is the next step?

Be sure to follow your baby's doctor's instructions for getting a new specimen for further testing as soon as you receive them. In the meantime, before the results of the additional test(s) are available, your doctor will talk with you about your baby's current health and will need to examine your baby. CAH is a rare disease involving the endocrine system, so your baby's doctor might consult with or refer you to a pediatric endocrinologist.

? What exactly is CAH?

CAH is caused by a missing enzyme. When this enzyme is missing, the adrenal glands produce too much of some hormones and not enough of others.

The salt-retaining hormone is one that is under-produced. Therefore babies with this condition are at risk of a serious sudden "salt-wasting" episode that can be fatal. A baby with CAH may appear normal at birth. Be sure to call your baby's doctor if any of these symptoms appear: a lot of vomiting, poor weight gain or if your baby looks ill.

There can also be other forms of CAH with less serious but still important effects. Children will usually develop normally if treatment begins in the first weeks of life. Finding out early if your baby has CAH, means you and your baby's doctor can take steps to prevent these effects.

👣 What can I do to prevent these effects?

Your baby needs to see the doctor right away. Secondly, the most important thing that you can do is to be sure your baby goes in for a new specimen to be collected and tested as soon as possible. If it turns out your baby does have CAH, there are very effective treatments available, and a pediatric endocrinologist will be able to help you manage this through monitoring the blood levels and adjusting the medication to help normalize hormone levels.

You have probably already done this, but now is also a good time to be sure your baby is covered by your health insurance provider.

NO treatment should be done unless a physician recommends it. Medications to treat CAH should NOT be started before collecting the specimen for the confirmatory test(s).

Remember, this could be a false alarm, but if it turns out your baby does have CAH, the treatment is very effective.

For more information talk with your baby's doctor, or you can contact the Nebraska Newborn Screening Program at (402) 471-0374 for information on Pediatric Endocrinologists available in Nebraska. A good web site for more information about CAH is at www.caresfoundation.org or at www.magicfoundation.org